SI gene

sucrase-isomaltase

Normal Function

The *SI* gene provides instructions for producing the enzyme sucrase-isomaltase. This enzyme is found in the intestinal tract, where it is involved in breaking down the sugars sucrose (a sugar found in fruits, and also known as table sugar) and maltose (the sugar found in grains). Sucrose and maltose are called disaccharides because they are each made up of two simple sugar molecules. Disaccharides must be broken down into simple sugar molecules to be digested properly.

The sucrase-isomaltase enzyme is found on the surface of the intestinal epithelial cells, which are cells that line the walls of the intestine. These cells have fingerlike projections called microvilli that absorb nutrients from food as it passes through the intestine. Based on their appearance, groups of these microvilli are known collectively as the brush border. The role of the sucrase-isomaltase enzyme is to break down sucrose and maltose into simple sugars so that they can be absorbed by microvilli into intestinal epithelial cells.

Health Conditions Related to Genetic Changes

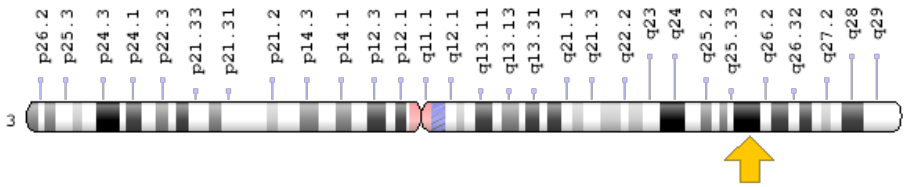
congenital sucrase-isomaltase deficiency

At least 10 mutations in the *SI* gene have been found to cause congenital sucrase-isomaltase deficiency. These mutations disrupt the folding and processing of the sucrose-isomaltase enzyme, transportation of the enzyme within the intestinal epithelial cells, the orientation of the enzyme to the cell surface, or its normal functioning. An impairment in any of these cell processes results in a sucrase-isomaltase enzyme that cannot effectively break down sucrose, maltose, or other compounds made from these sugar molecules (carbohydrates). The inability to digest these sugars causes the intestinal discomfort seen in people with congenital sucrase-isomaltase deficiency.

Chromosomal Location

Cytogenetic Location: 3q26.1, which is the long (q) arm of chromosome 3 at position 26.1

Molecular Location: base pairs 164,978,898 to 165,078,495 on chromosome 3 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- MGC131621
- MGC131622
- Oligosaccharide alpha-1,6-glucosidase
- sucrase-isomaltase (alpha-glucosidase)
- SUIS_HUMAN

Additional Information & Resources

Educational Resources

- Biochemistry (fifith edition, 2002): Sucrose, Lactose, and Maltose Are the Common Disaccharides
  https://www.ncbi.nlm.nih.gov/books/NBK22396/#A1519
- Molecular Cell Biology (fourth edition, 2000): The Intestinal Epithelium Is Highly Polarized
  https://www.ncbi.nlm.nih.gov/books/NBK21502/#A4119
- The Cell: A Molecular Approach (second edition, 2000): Glucose transport by intestinal epithelial cells
  https://www.ncbi.nlm.nih.gov/books/NBK9847/figure/A2010/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SI%5BTIAB%5D%29+OR+%28sucrase-isomaltase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D

OMIM

- SUCRASE-ISOMALTASE
  http://omim.org/entry/609845

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SI.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SI%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10856
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/6476
- UniProt
  http://www.uniprot.org/uniprot/P14410

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16543230
- Ritz V, Alfalah M, Zimmer KP, Schmitz J, Jacob R, Naim HY. Congenital sucrase-isomaltase deficiency because of an accumulation of the mutant enzyme in the endoplasmic reticulum. Gastroenterology. 2003 Dec;125(6):1678-85.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14724820

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16329100

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